KLLN gene

killin, p53-regulated DNA replication inhibitor

Normal Function

The *KLLN* gene provides instructions for making a protein called killin. The activity of the *KLLN* gene is controlled by a protein called p53 (which is produced from the *TP53* gene). Little is known about the function of killin, although it is thought to trigger cells to self-destruct (undergo apoptosis) when they are damaged or no longer needed. In this way, killin helps to prevent abnormal cells from growing and dividing unchecked to form tumors. Based on this role, killin is thought to be a tumor suppressor.

Health Conditions Related to Genetic Changes

Cowden syndrome

Some cases of Cowden syndrome and a similar condition called Cowden-like syndrome result from a change involving the *KLLN* gene. These conditions are characterized by multiple tumor-like growths called hamartomas and an increased risk of developing certain cancers. When Cowden syndrome and Cowden-like syndrome are caused by *KLLN* gene mutations, the conditions are associated with a particularly high risk of developing breast and kidney cancers.

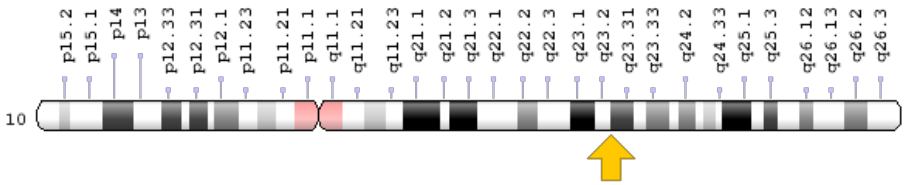
The genetic change associated with these conditions is known as promoter hypermethylation. The promoter is a region of DNA near the gene that controls gene activity (expression). Hypermethylation occurs when too many small molecules called methyl groups are attached to the promoter region. The extra methyl groups reduce the expression of the *KLLN* gene, which means that less killin is produced. A reduced amount of killin may allow abnormal cells to survive and proliferate inappropriately, which can lead to the formation of tumors.

The promoter region of the *KLLN* gene is shared with another gene, *PTEN*. The single promoter controls the expression of both genes. However, it appears that promoter hypermethylation only affects the expression of the *KLLN* gene; people with this type of genetic change have normal expression of the *PTEN* gene. Other types of mutations in the *PTEN* gene can cause Cowden syndrome and Cowden-like syndrome.

Chromosomal Location

Cytogenetic Location: 10q23.31, which is the long (q) arm of chromosome 10 at position 23.31

Molecular Location: base pairs 87,859,161 to 87,863,437 on chromosome 10 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KILIN_HUMAN
- killin

Additional Information & Resources

GeneReviews

- PTEN Hamartoma Tumor Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1488

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28KLLN%5BTIAB%5D%29+OR+%28killin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- KILLIN
  http://omim.org/entry/612105

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/KLLNID52121ch10q23.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KLLN%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=37212
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/100144748
- UniProt
  http://www.uniprot.org/uniprot/B2CW77

Sources for This Summary

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